Michigan Newborn Screening Program

November 1, 2005

Newborn Screening Goals

- Quick identification of newborns with rare, serious, but treatable disorders
- Early diagnosis and treatment of affected infants resulting in normal growth and development
- Reduction of significant human and financial costs for families and society

Criteria for Disorder

- Disorder is serious
- Reliable screening test in the newborn period
- Reasonable cost
- Effective treatment
- Medical facilities to confirm the diagnosis and provide treatment

Newborn Screening Law

MCL 333.5431

- No informed consent required
- Department may require that the tests be performed by the department
- Violation is a misdemeanor
- Fee for testing
- Annual increase in fee based on the CPI
- Allows blood specimens to be used for medical research during the retention period
- Parents can request second specimen to keep

Michigan Newborn Screening History

- 1965
  - Phenylketonuria
- 1977
  - Congenital Hypothyroidism
- 1984
  - Galactosemia
- 1987
  - Biotinidase Deficiency
  - Maple Syrup Urine Disease (MSUD)
  - Hemoglobinopathies
- 1993
  - Congenital Adrenal Hyperplasia (CAH)
- 2003
  - Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency (MCAD)
- 2004
  - Citrullinemia
  - Homocystinuria
  - Argininosuccinic Aciduria (ASA)
- 2005
  - Expanded Screen Pilot

Phenylketonuria

- Michigan Incidence: 1: 8,801
- Goal of PKU screening
  - Prevention of mental retardation
- Treatment
  - Lifelong diet management
    - Restriction of natural protein
    - Use of special formula
    - Use of modified low protein food products

Congenital Hypothyroidism

- Michigan Incidence 1: 1,942
- Goal of Screening
  - Prevent mental retardation and poor growth and development
- Treatment
  - Daily oral thyroid replacement

Galactosemia

- Michigan Incidence 1:41,227
- Goal of screening
  - Prompt identification and treatment to prevent neurological impairment and death
- Treatment
  - Immediate change to soy formula and lifelong exclusion of galactose from the diet

Maple Syrup Urine Disease (MSUD)

- Michigan Incidence 1: 234,992
- Goal of screening
  - Quickly identify affected infants to prevent neurological impairment and death
- Treatment
  - Strict diet limiting the intake of branched-chain amino acids and use of MSUD formula
  - Specially prepared branched-chain free TPN is used for acutely ill infants and children

Biotinidase Deficiency

- Michigan Incidence 1: 27,325
  - 2/3 Partial (10-30% enzyme activity)
  - 1/3 Profound (<10% enzyme activity
- Goal of Screening
  - Prevent neurological damage and death
- Treatment
  - Daily oral biotin supplements

Hemoglobinopathies (Sickle Cell Diseases)

- Michigan Incidence 1:1,956 Overall
  1: 600 African American newborns
  - Also seen in individuals of Mediterranean, Indian and Middle Eastern heritage
- Goal of Screening
  - Prevent death from pneumococcal sepsis
- Treatment
  - Penicillin prophylaxis until six years of age

Congenital Adrenal Hyperplasia (CAH)

- Michigan Incidence 1: 17,716
- Goal of Screening
  - Rapidly identify affected infants in order to prevent death from adrenal crisis or shock, and incorrect sex assignment in female newborns
- Treatment
  - Glucocorticoid , salt retaining hormones and surgical correction of ambiguous genitalia

Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)

- Michigan incidence ~1:26,205
- Goal of Screening
  - Prevention of hypoglycemia that could lead to coma, encephalopathy, liver failure or death
- Treatment
  - Frequent feedings are instituted to avoid fasting
  - Low-fat/high-carbohydrate diet and supplemental carnitine

Arginosuccinic Aciduria (ASA)

- Michigan incidence not yet determined
- Goal of Screening
  - Prevention of neurological damage and death
- Treatment
  - High caloric, protein restrictive diet
  - Sodium Benzoate and Sodium Phenylacetete supplementation may be used

Citrullinemia

- Michigan incidence not yet determined
- Goal of Screening
  - Prompt identification and treatment to prevent neurological damage and death
- Treatment
  - High caloric, protein restricted diet
  - Sodium Benzoate and Sodium Phenylacetate supplements may be used
  - Medications to remove waste products from the blood
  - Dialysis may be necessary

Homocystinuria

- Michigan incidence not yet determined
- Goal of Screening
  - Prevention of mental retardation, seizures, optical lens dislocation, osteoporosis, scoliosis, and/or thrombi formation
- Treatment
  - Lifelong strict diet limiting the intake of methionine through the use of special infant formulas.
  - Cystine and vitamin B6 supplements may be used

Michigan Newborn Screening Facts

- Approximately 128,000 births per year
- Approximately 1,200 non-hospital births/year
- 99 birthing hospitals or hospitals with an NICU

Children Identified in 2004

- PKU - 12
  - 8 diet treated
- Hypothyroidism – 66
- Galactosemia - 2
- MSUD - 2
- Biotinidase – 23
  - 1 Profound
  - 22 Partial
- Sickle Cell Diseases - 73
  - FS- 37
  - FSA- 15
  - FSC- 19
  - FSV- 2
- CAH - 8
- MCAD – 6

Original Specimens tested in NBS lab: 127,572

Total Infants Identified: 192

Combined Incidence: 1 per 664 births

Program Funding

- Providers are charged for first sample "blue" cards ($56.83)
- All repeat test "pink" cards supplied at no additional cost
- Funding supports
  - Laboratory
  - Follow Up program
  - Medical Management programs

Medical Management

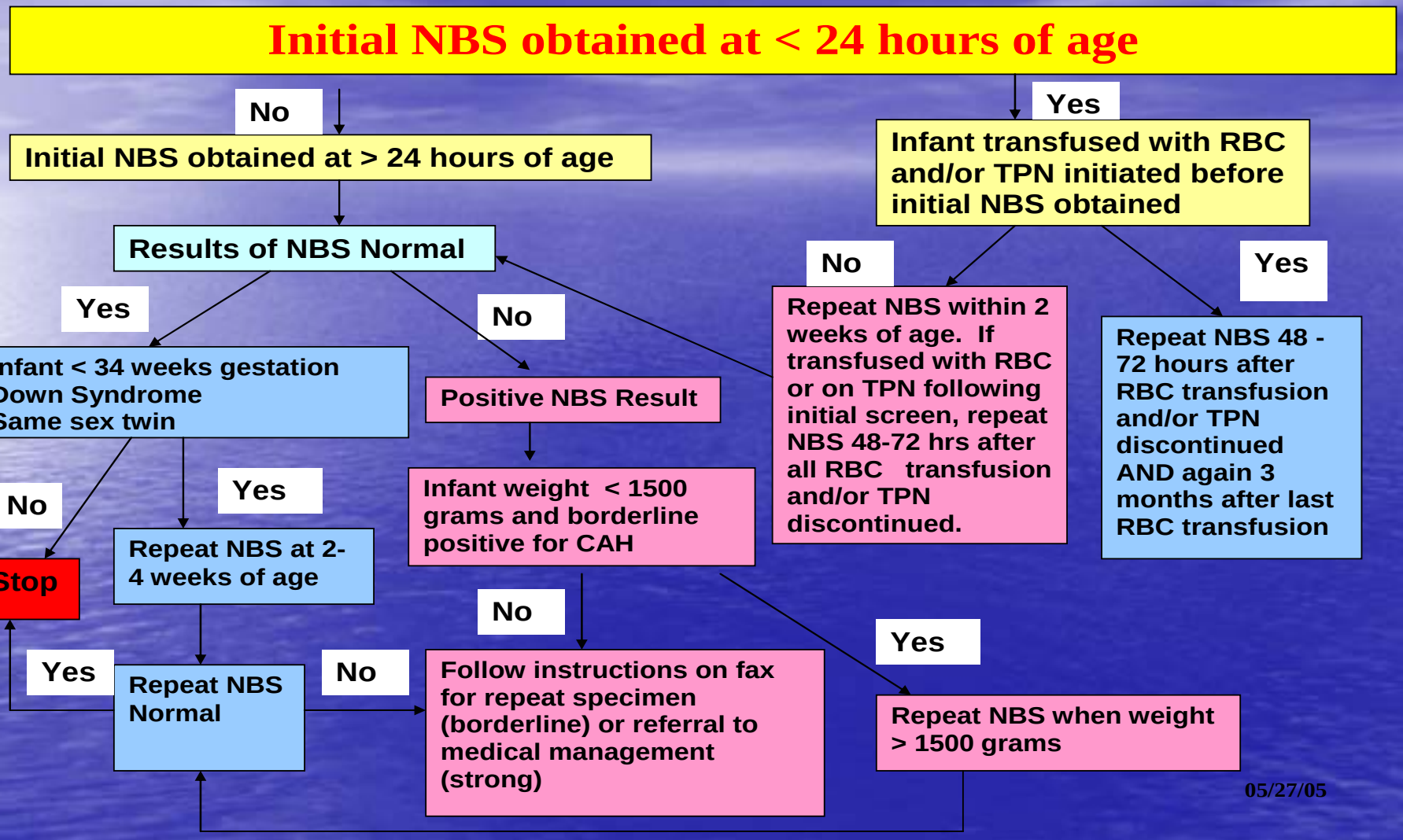
- Children's Hospital of Michigan Metabolic Clinic (CHMMC) 313-745-4513
- Endocrine Follow up program (EFUP) at University of Michigan (734) 647-8938
- Sickle Cell Disease Association of America (SCDAA), Detroit Chapter
  (313) 864-4406

Screening Recommendations

- Obtain initial screen at 24-36 hours of age
- Always test before discharge or transfer regardless of age
  - Follow directions on Early Specimen letter from the State Laboratory regarding repeat specimens
- Always test before transfusion of red blood cells or the administration of TPN

NICU Screening Algorithm

DRAFT



Completing the Card

- Accurate information is vital
  - Identification /location of infants for follow up of abnormal results
    - Inaccurate information could cause a life-threatening delay for affected infants
  - Age (in hours) and weight (in grams) are critical to provide accurate screening results

Completing the Card

- Remove hearing slip before blood collection
- All information should be accurate, legible and complete

The sample submitter is legally responsible for the accuracy and completeness of the information on the newborn screening card

Completing the Card

Press Firmly with Pen

Critical Demographic Information

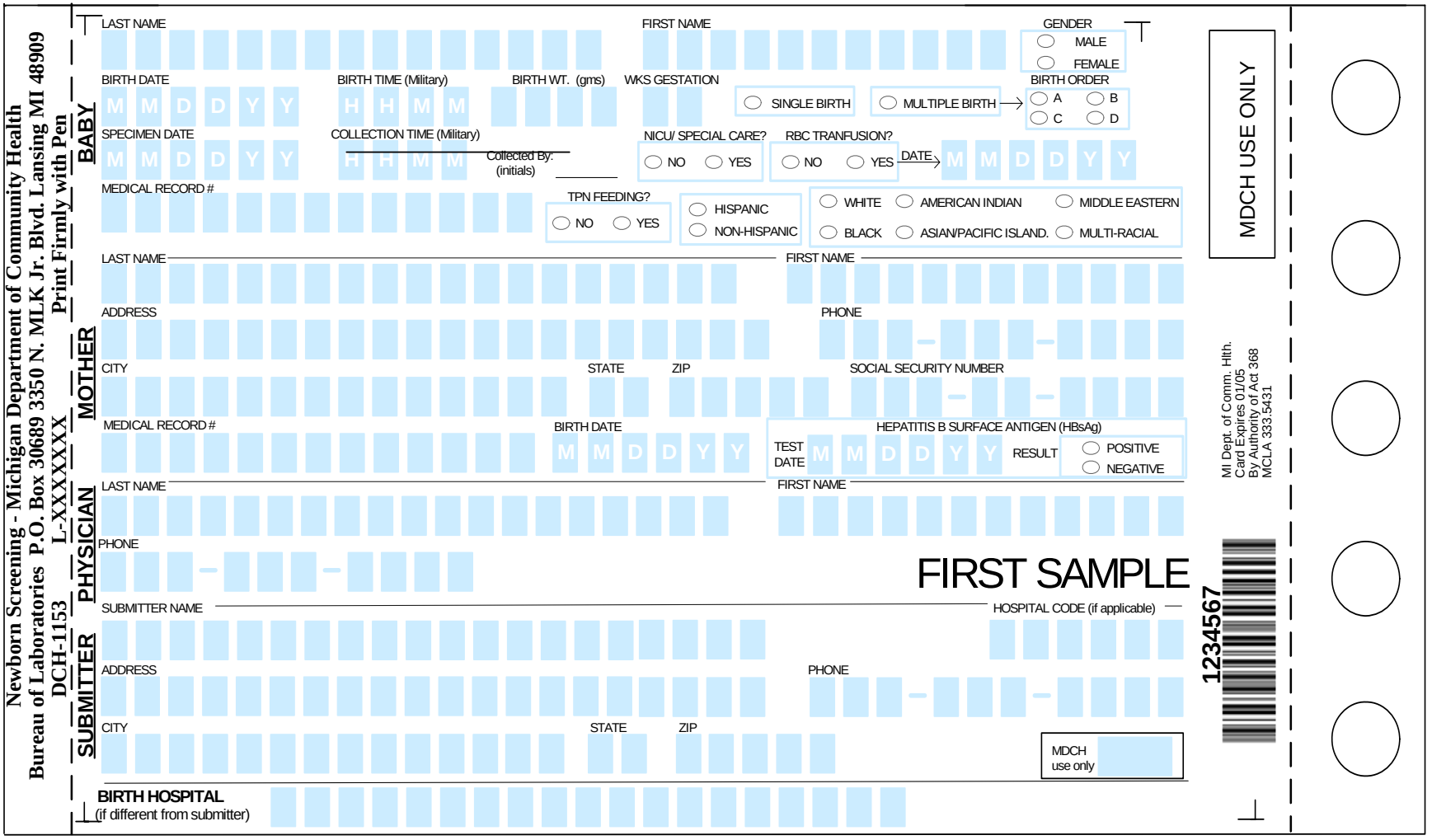
Baby's last name
Mother's first and last name
Mother's social security #
Date and time of birth
Birth weight in grams
Gestational age
Date and time of specimen collection
Date of transfusion (red blood cells)
TPN feeding Yes or No

Screening Cards

Features of the card

- Submitter copy
- Date/time of birth and date/time of specimen collection
- Space for initials of person collecting specimen
- Overlay flap to protect filter paper before collection

Revised Newborn Screening Card



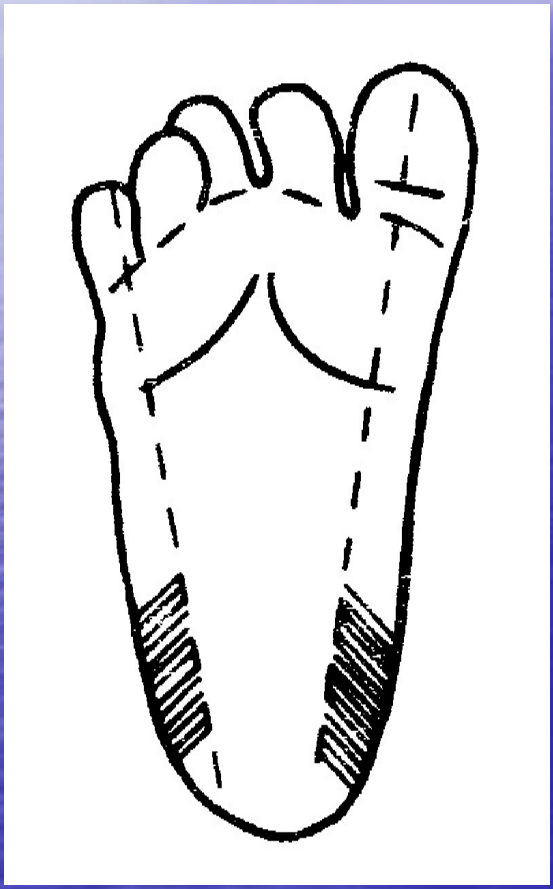
Collection Procedure

- The MDCH follows the recommendations of the National Committee for Clinical Laboratory Standards (NCCLS) for collecting an acceptable specimen
  - "Blood Collection on Filter Paper for Neonatal Screening Programs; Approved Standard- Fourth Edition" (July 2003)

Collection Procedure

- Use capillary blood from heelstick
- Fill in all five circles
- Apply blood to **only one side** of the filter paper
- Dry Flat at least 3 hours
- Mail to state laboratory within 24 hours of collection

Collection Procedure



- Warm foot for 3 – 5 minutes to increase blood flow
- Cleanse site with alcohol prep
- Air dry or wipe dry with sterile gauze pad
- See picture for recommended puncture site

Collection Procedure

- Puncture heel with lancet of no more than 2.0 mm in depth
- Wipe away first drop of blood
- Apply gentle pressure to allow a large drop of blood to form
- Lightly touch filter paper to large drop of blood
- Allow blood to soak through to completely fill the circle

Indwelling Line Procedure

- Do not draw from intravenous lines where TPN or blood is being infused
- For other types of IV fluid, make sure the line has been thoroughly flushed
- Avoid the use of syringes with additives
- Spot the card immediately after collection

Feeding/Antibiotic/TPN Issues

- Feeding status is no longer a factor with technologies currently used in the MDCH Newborn Screening Laboratory
- Antibiotics have no effect on the NBS with current laboratory testing methods
- TPN may cause false positive screening tests for PKU and MSUD
  - Screening results should return to normal in unaffected babies when TPN discontinued

Things to Avoid

DO NOT

- Apply blood to both sides of the filter paper
- Apply "layers" of blood onto the same circle
- Apply excessive amounts of blood (circles should not touch one another)
- Allow filter paper to come in contact with other substances

Satisfactory Specimens

Front

Back

Note the even penetration of blood that indicates a single large drop of blood was applied to the filter paper. The circle should look the same when viewing the card front or back.

Valid Specimen

Printed with permission from Schleicher & Schuell and the New York State Department of Health

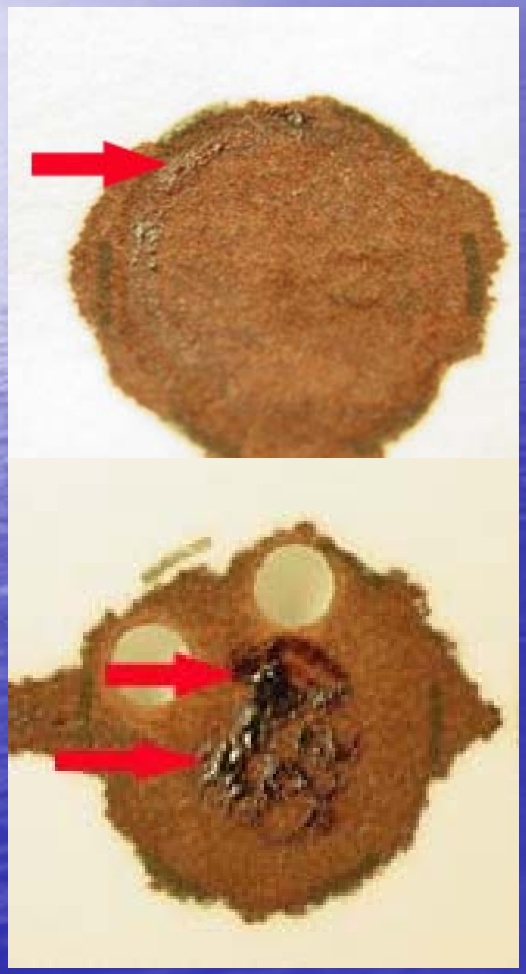
Allow sufficient amount of blood to soak through to completely fill the circle

Unsatisfactory Specimens

- Failure to follow correct procedures will likely result in an unsatisfactory sample.
  - These samples will not yield reliable results
  - repeat test will be necessary

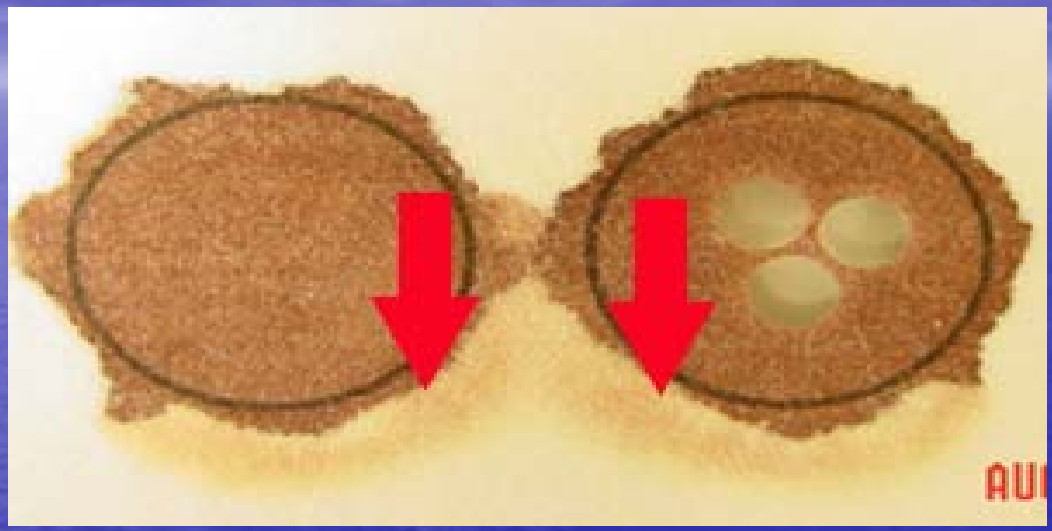
The delay caused by an unsatisfactory sample could be life-threatening to an affected child.

Clotted Specimens



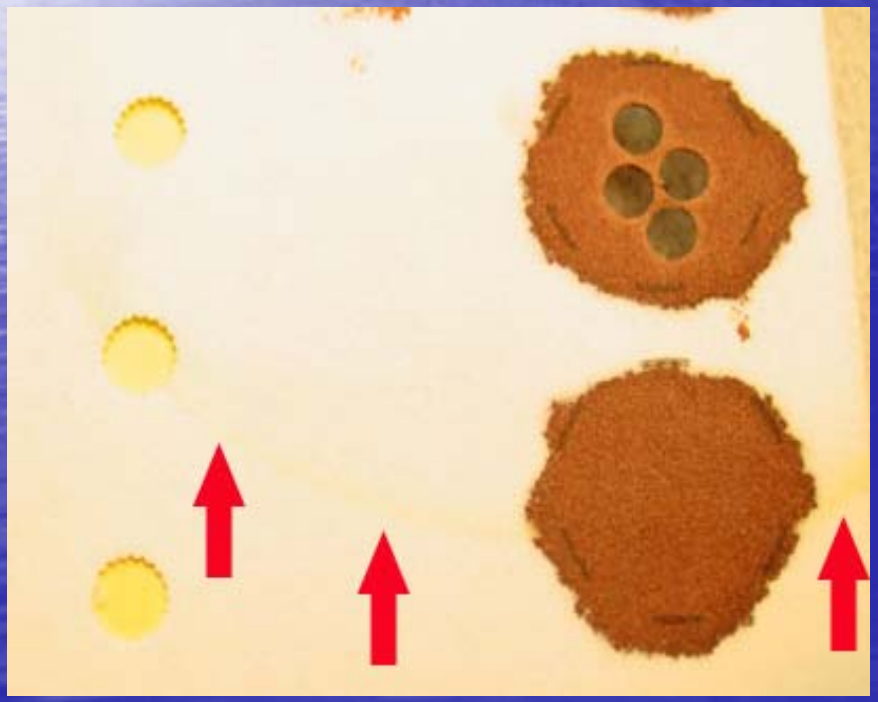
- Causes
  - Waiting too long to apply blood to the filter paper
  - Improper use of capillary tubes
  - Syringe used for blood collection
- Prevention
  - Warm heel to assure good blood flow
  - Follow MDCH guidelines for capillary tube collection
  - Avoid application of excessive amounts of blood

Serum Rings



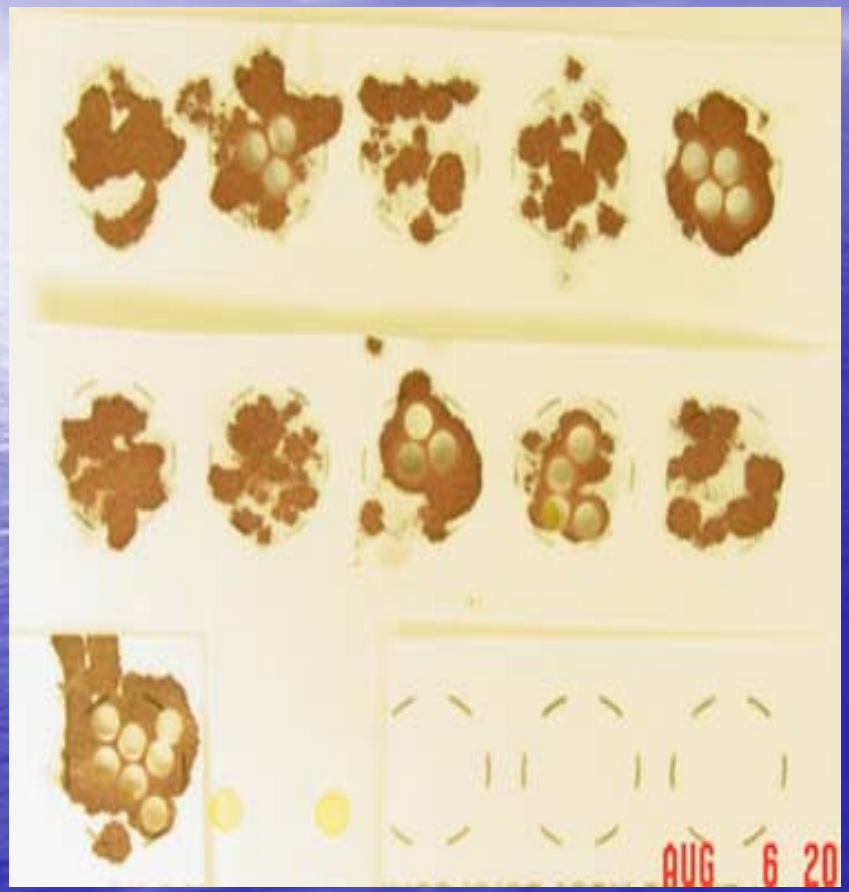
- Causes
  - Milking or squeezing around puncture site
  - Improper drying of specimen (up on side)
- Prevention
  - Wipe away first drop of blood (contains tissue fluid)
  - Protect from excessive heat
  - Dry in horizontal (flat) position

Contaminated Specimens



- Causes
  - something spilled on the filter paper or it was set on a wet surface prior to or after the application of blood
- Prevention
  - protect the filter paper from coming in contact with hands or other substances before and after blood collection..

Insufficient Specimens



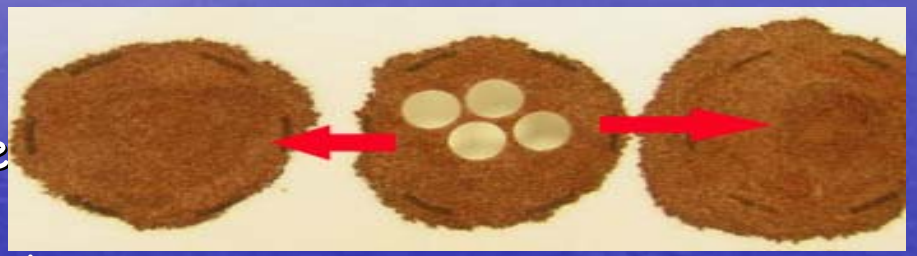
- Causes
  - Poor blood flow
  - Lancet did not make good puncture
  - All circles not filled
  - Entire circle not filled
  - Blood did not soak through filter paper
- Prevention
  - Warm heel before blood collection
  - Use lancet that makes a wound 2.0mm deep
  - Fill all circles

Diluted Specimens

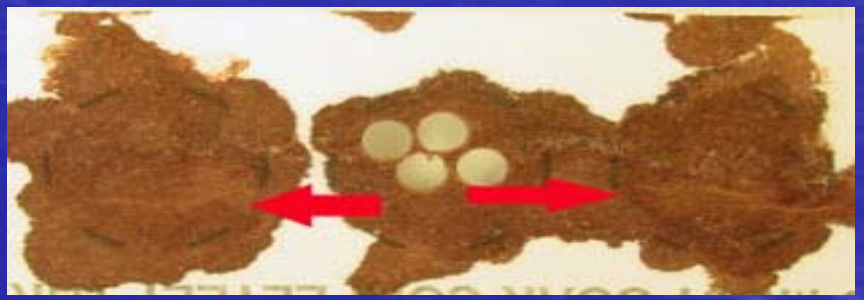
- Causes
  - Blood collected from an indwelling line that was not cleared prior to obtaining the specimen
- Prevention
  - Draw appropriate amount of blood (e.g.2 to 2.5cc) from the line before sample is obtained.

Layered Specimens

- Causes
  - Multiple applications of blood to the same circle
  - Blood applied to both side of the filter paper
  - Unevenly distributed blood
  - Circles of blood touch or overlap



- Prevention
  - Allow one large drop to soak through and fill the entire circle
  - Apply blood to only one side
  - Apply blood in circle only



Scratched or Damaged Specimens

- Causes
  - Improper use of capillary tubes and other collection devices
  - Pressing the heel against the filter paper when obtaining the screening sample.
- Prevention
  - Follow MDCH guidelines for capillary tube collection
  - Avoid touching the filter paper with heel or collection device

Over Saturated Specimens

- Causes
  - Application of excessive amounts of blood to the filter paper
  - Circles should not touch
- Prevention
  - Apply blood in the preprinted circles only

Stuck to Backing

- Causes
  - Back of card not folded away during drying of blood spots
- Prevention
  - . Do not allow the flap/backing to come in contact with the blood spots until dry.

Defective Form

This specimen was collected on a defective or damaged form. Note the areas indicated by the arrows where the blood did not adhere to the form. When this happens, the process should be repeated on a new form.

Drying/Mailing Instructions

- Air dry specimen FLAT for at least 3 hours
  - Keep away from heat and direct sunlight
- Mail specimens within 24 hours of collection
  - Do not hold specimens for bulk mailing
  - Pre-addressed envelopes are available for prompt mailing

Laboratory Procedures

- Specimens are tested the day they are received
  - Up to 700 Specimens each day
- Preliminary results same day for life threatening disorders
- Unsatisfactory specimens reported the same day they are received
  - All tested and positives reported

Follow Up

- Positive results are followed up with a repeat screening test or prompt referral to medical management
- Telephone/fax notification with instructions to local physician
- Negative results sent to hospital
  - Hospital forwards to local physician

Supplemental Screening

- Testing for additional disorders beyond the Michigan eleven
- Parents can obtain kit from supplemental screening lab (Pediatrix, Baylor, Mayo, University of CO)
- Blood is collected at same time as state test
- No follow-up component

Issues of Concern

- Unscreened Infants
  - Match hearing screens to metabolic
  - Match NBS to birth certificates
  - Transfers and early discharge - Birth hospital is responsible for obtaining the initial screen

Do not assume no news is good news, it may mean not done!

Issues of Concern

- "Late" Specimens: > 6 days from birth date to punch date in NBS Lab
  - Causes
    - Specimens drawn at > 36 hours of age
    - Batching of specimens
    - Mailroom delays
    - Weekends

Strategies for Improvement

- Quarterly Quality Assurance Reports
- "Newborn Screening Update"
- Site Visits
- Web based educational program
  - www.training.mihealth.org
  - Web site
  - www.mi.gov/newbornscreening

Replacement Blue Cards

OOPS!

If blue cards (initial test) are "damaged" by:

- Improper specimen collection
- Mishandling
- Errors in recording patient or hospital information

Replacement cards can be obtained at no charge.

Replacement Procedure

- **Remove the filter paper portion if it contains blood (very important)**
- Complete "Newborn Screening Card Replacement Form"
  - Forms available from NBS Accountant, Valerie Klasko at (517) 241-5583

Replacement Procedure

- Indicate clearly on card reason for return
- Mail with submitter's return address to;

  Michigan Department of Community Health
  Attention: Newborn Screening
  Lewis Cass Building 4th Floor
  320 South Walnut
  Lansing, MI 48913

It's Not Just PKU!

The current newborn screening panel tests for 11 disorders. To avoid confusion, it is important to use correct terminology when referring to newborn screening tests.

Please make every effort to call the test "Newborn Screen" rather than "PKU"

Michigan Newborn Screening Program Brochure

- Newborn Screening brochures are available to assist in discussing the NBS screening process with parents and prospective parents
- Brochures contain information on the 11 disorders currently being screened
- Brochures are available from NBS Accountant, Valerie Klasko (517) 241-5583

Free Online Program for Newborn Screening Education

- Go to www.training.mihealth.org
- Follow the directions to log in – free
- Select Newborn Screening
- Take the course

You can take the whole course or one part at a time. Average completion time is one hour or less. A certificate is provided if a passing score is received on the final quiz.

Contact Information

Newborn Screening Program
Telephone: (517) 335-9205
Fax: (517) 335-9419
Email: mdch-newbornscreening@michigan.gov
Website: www.michigan.gov/newbornscreening

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